



Overview

Points To Remember About Osteogenesis Imperfecta (OI)

- OI is a genetic disease that causes bones to weaken and break easily.
- There are several types of OI. The type of OI you have affects the symptoms you have.
- Symptoms of OI range mild to severe. All people with OI have weak bones that break easily.
- OI is usually managed by a team of doctors.
- There are several treatments for OI, including medicines and surgery. Treatments may help prevent or control symptoms of OI.
- There are steps you can take to stay healthy and help prevent broken bones.
- There is no way to prevent OI.

Osteogenesis imperfecta (OI), also called brittle bone disease, is a genetic disease in which the bones of the body break easily, often with no obvious cause.

What happens in OI?

OI is caused by a genetic defect in one of the genes that carry instruction for making collagen. Collagen is a material in bones that helps make them strong. This defect may result in collagen that does not work properly or not enough collagen. Either leads to weak bones that break easily.

Who Gets

About 20,000 to 50,000 people in the United States have OI. Though anyone can be born with OI, people with a family history of the disease are at greater risk of inheriting the disease

through an abnormal gene that is passed on from one or both parents. Genetic counselors can help you better understand the genetics of OI. (For more information, see [Causes of OI](#)).

Types

There are eight types of OI. The type of OI a person has affects how mild or severe the symptoms of the disease is. Type 1 is the mildest and most common form of OI. Type 2 is the most severe form of OI. Other types of OI have symptoms that fall between Type 1 and Type 2; types 3, 7, and 8 have less severe symptoms, while types 4, 5, and 6 have more moderate symptoms.

Type 1 OI

- Bones likely to break, with most broken bones occurring before puberty.
- Normal or near-normal stature.
- Loose joints and muscle weakness.
- Blue, purple, or gray tint to sclera (whites of the eyes).
- Triangular face.
- Curved spine.
- No or little minimal bone deformity.
- Possible brittle teeth.
- Possible hearing loss, often beginning in early twenties or thirties.
- Normal collagen structure, but less than normal amount.

Type 2 OI

- Frequently causes death at birth or shortly after, because of lung problems.
- Numerous broken bones.
- Severe bone deformity.
- Small stature.
- Underdeveloped lungs.
- Blue, purple, or gray tinted sclera.
- Improperly formed collagen.

Type 3 OI

- Most severe type among those who survive the neonatal period.
- Easily broken bones. (Broken bones are often present at birth, and x-rays may reveal healed bone breaks that occurred before birth.)
- Small stature.
- Blue, purple, or gray tinted sclera.
- Loose joints.
- Poor muscle development in arms and legs.
- Barrel-shaped rib cage.
- Triangular face.

- Curved spine.
- Possible lung problems.
- Often severe bone deformity.
- Possible brittle teeth.
- Possible hearing loss.
- Improperly formed collagen.

Type 4 OI

- Between Type I and Type III OI in severity.
- Bones break easily, with most broken bones occurring before puberty.
- Smaller than average stature.
- Sclera normal in color.
- Mild to moderate bone deformity.
- Tendency toward curved spine.
- Barrel-shaped rib cage.
- Triangular face.
- Possible brittle teeth.
- Possible hearing loss.
- Improperly formed collagen.

Type 5 OI

- Clinically similar to Type IV OI in appearance and symptoms.
- A dense band seen on x-rays by the growth plate of the long bones.
- Unusually large calluses, called hypertrophic calluses, at the sites of fractures or surgical procedures. (A callus is an area of new bone that is laid down at the fracture site as part of the healing process.)
- Calcification of the membrane between the radius and ulna (the bones of the forearm), which results in restricted arm movement.
- Sclera normal in color.
- Normal teeth.
- “Mesh-like” appearance to bone when viewed under the microscope.

Type 6 OI

- Similar to Type IV OI in appearance and symptoms, but is extremely rare.
- Slightly elevated level of an enzyme linked to bone formation called alkaline phosphatase, which can be determined by a blood test.
- “Fish-scale” appearance to bone when viewed under the microscope.
- Diagnosed by bone biopsy.

Type 7 OI

- Resembles Type IV OI in many aspects of appearance and symptoms in the first described cases.
- In other instances, similar appearance and symptoms to Type II OI, except infants had white sclera, a small head, and a round face.

- Small stature.
- Short humerus (arm bone) and short femur (upper leg bone).
- A deformed hip joint in which the neck of the femur is bent downward; this condition is called coxa vera.

Type 8 OI

- Resembles Type II or Type III OI in appearance and symptoms, except infants have white sclera.
- Severe growth deficiency.
- Bones lack important minerals that keep them strong.

Symptoms

All people with OI have weak, brittle bones. Some people with OI may have a few broken bones over their lifetime. Others may have hundreds of broken bones in their lifetime, including broken bones that occur before birth.

People with OI may have others symptoms, which can range from mild to severe and vary from person to person. These include:

- Malformed bones.
- Small stature.
- Skin the bruises easily.
- Loose joints.
- Weak muscles.
- Whites of the eyes that look blue, purple, or gray.
- A face shaped like a triangle.
- A rib cage shaped like a barrel.
- A curved spine.
- Brittle teeth.
- Hearing loss.
- Breathing problems.

Causes

OI is caused by an abnormal gene. Genes carry information that determine which features are passed to you from your parents. We have two copies of most of our genes, one from each parents.

People with OI have an abnormal gene that helps make collagen, which makes bones strong. People with OI do not have enough collagen. Or, the collagen does not work properly. This causes weak bones that break easily.

Most people with OI inherit this gene from one parent. Others inherit it from both parents. Parents do not have to have OI to pass on the gene that causes it. Sometimes, neither parent passes on the gene. Instead, the gene stops working properly on its own before the child is born.

Dominant OI

Most people with OI have what is called a dominant form of OI. This means they inherited one normal copy and one abnormal copy of one of the genes that cause OI. The abnormal copy of the gene is stronger than or “dominant” over the normal copy of the gene. This causes a person to be affected by OI. A person with OI caused by a dominant mutation has a 50 percent chance (1 in 2) of passing on the disorder to each of his or her children. Some children who have the dominant form of OI inherit the one of the genes that cause OI from a parent. Other children are born with the dominant form of OI even though there is no family history of the disorder.

Recessive OI

Other people with OI have a recessive form of the disease. People with recessive OI have parents who do not have OI but who both have an abnormal gene that causes OI. The parents of a child with recessive OI have a 25-percent chance (1 in 4) per pregnancy of having another child with OI. Siblings of a person with recessive OI have a 50-percent chance (1 in 2) of having an abnormal gene that causes OI. If one parent has OI because of a recessive mutation, their children will have an abnormal gene that causes OI, but will not necessarily have OI.

Genetic counselors can help people with OI and their family members understand the genetics of OI.

Diagnosis

There is no single test to detect OI. Doctors look at several factors to diagnose OI. These include:

- Family history.
- Medical history.
- Results from a physical exam.
- X-rays.

OI can also be diagnosed through a skin biopsy. A skin biopsy is a surgical procedure in which a doctor removes a small piece of skin. Then, a doctor looks at it under a microscope to see whether there are any collagen abnormalities. In addition, doctors can also diagnose OI through a blood test that detects the abnormal gene that causes OI. These tests detect OI in 9 out of 10

people who have it.

Treatment

Although there are no medications approved by the U.S. Food and Drug Administration to treat OI, your doctor may recommend a therapy approved for a related condition. The goal of treatments is to prevent or control symptoms of OI, developing bone mass and muscle strength, and maximizing a person's ability to be independent. These treatments include:

Medicines

Your doctor may prescribe:

- **Bone strengthening medicines** to slow bone loss and reduce the frequency and seriousness of broken bones.
- **Pain medicines** to treat pain caused by broken bones.
- **Other treatments**, such as growth hormone treatment, gene therapies, and an injected drug called teriparatide (for adults only), which are under study.

Broken Bone Care

Your doctor may cast, splint, or brace a broken bone to help it heal correctly.

Surgery

Surgery is a common treatment option for people with OI. Surgery is often recommended by doctors to fix a broken bone, support or correct bones that are curved or bowed, or to support the spine.

Many children with OI have surgery in which a metal rod is placed into a bone. This is called rodding surgery. Rodding surgery is performed to support the bone and prevent the bone from breaking.

Mobility Aids

Using a mobility aid may help you safely perform daily activities and reduce injuries. Walkers, canes, crutches, or wheelchairs are commonly used by people who have OI.

Physical or Occupational Therapy

People with OI may benefit from physical or occupational therapy, which can help:

- Build muscle strength, which may help prevent broken bones.
- Learn how to avoid injuries.
- Safely perform activities of daily living.
- Recover from broken bones.

Dental Care

Some people with OI have brittle teeth that chip or crack easily. If you have brittle teeth, you may require special dental care.

Who Treats

People with OI usually require a health care team made up of several doctors and healthcare providers. Your health care team may include:

- Internists, who diagnose and treats adults.
- Pediatricians, who diagnose and treats children.
- Orthopaedists, who treat and perform surgery for bone and joint diseases.
- Occupational therapists, who teach how to safely perform activities of daily living.
- Physical therapists, who teach ways to build muscle strength and prevent broken bones. Physical therapists may also help you recover from broken bones.
- Nutritionists, who teach how to use diet to improve overall health and stay at a healthy weight.

Living With

Certain activities can help you stay healthy and prevent broken bones.

- Follow a nutritious diet.
- Exercise as much as possible. Regular physical activity can help strengthen muscles and bones. Swimming and water therapy are common choices for people with OI because exercising in water has little risk of broken bones. Talk with your doctor or physical therapist to discuss appropriate and safe exercise.
- Keep a healthy weight. Being overweight increases the risk for many health problems, such as diabetes and heart disease. Extra weight also adds stress to the bones, which is especially unhealthy for people with OI.
- Don't smoke. Smoking causes cancer, heart disease, and other serious health conditions. Smoking can also weaken bones.
- Do not drink a lot of alcohol or caffeine. They may weaken your bones.
- Do not take steroid medicines. They can weaken bones.

Research Progress

Research is underway to help people with OI, including:

- Genes that cause OI.
- Medications to help people with OI grow.
- Drugs to make bones stronger.
- Better devices to use in surgery.

Related Resources

U.S. Food and Drug Administration

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

[Drugs@FDA](https://www.accessdata.fda.gov/scripts/cder/daf) at <https://www.accessdata.fda.gov/scripts/cder/daf> [Drugs@FDA](https://www.accessdata.fda.gov/scripts/cder/daf) is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics

Website: <https://www.cdc.gov/nchs>

NIH Osteoporosis and Related Bone Diseases ~ National Resource Center

Website: <https://www.bones.nih.gov>

Osteogenesis Imperfecta Foundation

Website: <https://www.oif.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- [Asian Language Health Information](#)
- [Spanish Language Health Information](#)

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Related Information

[Osteogénesis Imperfecta: Esenciales: hojas informativas de fácil lectura](#)

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[Osteogenesis Imperfecta, Easy-to-Read Fast Facts](#)

[Osteogenesis Imperfecta Overview](#)

[OI Issues: Maintaining Health During the Adult Years](#)